

Figure 1: Testosterone biosynthetic pathway

CYP17A1

26 865 bp



	SNP29: -34T>C	SNP04: IVS1 +426G>A	SNP20: IVS1 +466G>A	SNP06: IVS1 -99C>T	SNP07: IVS2 +105A>C	SNP22: IVS2 -83C>T	SNP03: IVS5 +75C>G
Hap1	T	C	C	C	A	C	C
Hap2	C	T	.	T	C	.	G
Hap3	C	T	G
Hap4	Composite						

Frequency (all)		Frequency (EA)		Frequency (AA)	
Controls	Cases	Controls	Cases	Controls	Cases
547 (.57)	527 (.60)	494 (.57)	475 (.60)	50 (.66)	47 (.62)
259 (.27)	216 (.25)	253 (.29)	208 (.26)	5 (.07)	6 (.08)
91 (.09)	86 (.10)	85 (.10)	79 (.10)	3 (.04)	7 (.09)
63 (.07)	51 (.06)	42 (.05)	32 (.04)	18 (.24)	16 (.21)

Figure 2A

CYP3A4

37 073 bp



	SNP47: -1232C>T	SNP12: -747C>G	SNP11: -392A>G	SNP01: IVS7 +34T>G	SNP13: IVS7 -202C>T	SNP24: stop +766 delT	SNP25: stop +1454C>T	SNP05: stop +1639A>T	SNP15: stop +2204G>C
Hap1	C	C	A	T	C	T	C	A	G
Hap2	.	G
Hap3	T	G	.	T	C
Hap4	T	.	G	G	T	G	.	T	C
Hap5	Composite								

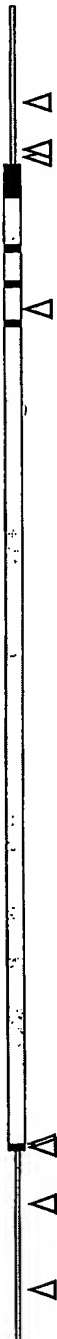
Frequency (all)		Frequency (EA)		Frequency (AA)	
Controls	Cases	Controls	Cases	Controls	Cases
651 (.68)	596 (.68)	629 (.72)	577 (.73)	16 (.21)	13 (.17)
65 (.07)	62 (.07)	64 (.07)	59 (.07)	1 (.01)	2 (.03)
63 (.07)	64 (.07)	59 (.07)	58 (.07)	4 (.05)	6 (.08)
45 (.05)	25 (.03)	34 (.04)	19 (.02)	10 (.13)	5 (.07)
136 (.14)	133 (.15)	88 (.10)	81 (.10)	45 (.59)	50 (.66)

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Figure 2B

SRD5A2

76 341 bp



SNP17: - 8029C>T
 SNP26: - 3001G>A
 SNP22: A49T (145G>A)
 SNP20: V89L (265G>C)
 SNP12: IVS2 + 626C>T
 SNP01: stop + 1552G>A
 SNP13: stop + 3059G>A
 SNP15: stop + 9301G>C

	Frequency (all)		Frequency (EA)		Frequency (AA)	
	Controls	Cases	Controls	Cases	Controls	Cases
Hap1	376 (.39)	348 (.40)	352 (.40)	321 (.40)	19 (.25)	20 (.26)
Hap2	218 (.23)	212 (.24)	206 (.24)	197 (.25)	11 (.14)	13 (.17)
Hap3	108 (.11)	84 (.10)	102 (.12)	81 (.10)	6 (.08)	3 (.04)
Hap4	73 (.08)	73 (.08)	70 (.08)	65 (.08)	-	7 (.09)
Hap5	41 (.04)	42 (.05)	41 (.05)	42 (.05)	-	-
Hap6	32 (.03)	27 (.03)	19 (.02)	20 (.03)	13 (.17)	7 (.09)
Hap7 Composite	112 (.12)	94 (.10)	84 (.10)	68 (.09)	27 (.36)	26 (.34)

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Figure 2C